



## nonbullous congenital ichthyosiform erythroderma

Nonbullous congenital ichthyosiform erythroderma (NBCIE) is a condition that mainly affects the skin. Some affected infants are born with a tight, clear sheath covering their skin called a collodion membrane. This membrane is usually shed during the first few weeks of life. Individuals with NBCIE have skin that is red (erythema) and covered with fine white scales. Some people with NBCIE have outward turning eyelids and lips, a thickening of the skin on the palms and soles of the feet (keratoderma), and nails that do not grow normally (nail dystrophy). Infants with NBCIE may develop infections, an excessive loss of fluids (dehydration), and respiratory problems early in life.

### Frequency

NBCIE is estimated to affect 1 in 200,000 to 300,000 individuals in the United States. This condition is more common in Norway, where an estimated 1 in 90,000 people are affected.

### Genetic Changes

Mutations in at least three genes can cause NBCIE. These genes provide instructions for making proteins that are found in the outermost layer of the skin (the epidermis). The epidermis forms a protective barrier between the body and its surrounding environment. The skin abnormalities associated with NBCIE disrupt this protective barrier, making it more difficult for affected infants to control water loss, regulate body temperature, and fight infections.

Mutations in the *ALOX12B* and *ALOXE3* genes are responsible for the majority of cases of NBCIE. Mutations in one other gene associated with this condition are found in only a small percentage of cases. In some people with NBCIE, the cause of the disorder is unknown. Researchers are looking for additional genes that are associated with NBCIE.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- collodion baby
- congenital nonbullous ichthyosiform erythroderma

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855792/>

### Other Diagnosis and Management Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST): Treatments  
[http://www.firstskinfoundation.org/content.cfm/category\\_id/0/page\\_id/830](http://www.firstskinfoundation.org/content.cfm/category_id/0/page_id/830)
- GeneReview: Autosomal Recessive Congenital Ichthyosis  
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Skin Conditions  
<https://medlineplus.gov/skinconditions.html>

### Genetic and Rare Diseases Information Center

- Nonbullous congenital ichthyosiform erythroderma  
<https://rarediseases.info.nih.gov/diseases/9736/nonbullous-congenital-ichthyosiform-erythroderma>

## Educational Resources

- Disease InfoSearch: Ichthyosiform erythroderma, nonbullous congenital  
<http://www.diseaseinfosearch.org/Ichthyosiform+erythroderma%2C+nonbullous+congenital/3710>
- MalaCards: ichthyosis, congenital, autosomal recessive 2  
[http://www.malacards.org/card/ichthyosis\\_congenital\\_autosomal\\_recessive\\_2](http://www.malacards.org/card/ichthyosis_congenital_autosomal_recessive_2)
- MalaCards: ichthyosis, congenital, autosomal recessive 3  
[http://www.malacards.org/card/ichthyosis\\_congenital\\_autosomal\\_recessive\\_3](http://www.malacards.org/card/ichthyosis_congenital_autosomal_recessive_3)
- Merck Manual Professional Version: Ichthyosis  
<http://www.merckmanuals.com/professional/dermatologic-disorders/cornification-disorders/ichthyosis>
- The Swedish Information Centre for Rare Diseases  
<http://www.socialstyrelsen.se/rarediseases/ichthyosis>

## Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases  
<http://www.climb.org.uk/>
- Foundation for Ichthyosis and Related Skin Types (FIRST): Congenital Ichthyosiform Erythroderma  
[http://www.firstskinfoundation.org/content.cfm/category\\_id/741/page\\_id/541](http://www.firstskinfoundation.org/content.cfm/category_id/741/page_id/541)
- National Organization for Rare Disorders (NORD): Ichthyosis  
<https://rarediseases.org/rare-diseases/ichthyosis/>
- University of Kansas Medical Center Resource List  
<http://www.kumc.edu/gec/support/ichthyos.html>

## GeneReviews

- Autosomal Recessive Congenital Ichthyosis  
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

## ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22nonbullous+congenital+ichthyosiform+erythroderma%22+OR+%22Congenital+Ichthyosiform+Erythrodermas%22+OR+%22Congenital+Ichthyosiform+Erythroderma%22+OR+%22Ichthyosiform+Erythroderma%2C+Congenital%22+OR+%22Ichthyosis%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28nonbullous+congenital+ichthyosiform+erythroderma%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 2  
<http://omim.org/entry/242100>

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